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Life-Threatening Splenic Hemorrhage in Two Patients With Gaucher Disease

Deborah L. Stone, Edward I. Ginns, Donna Krasnewich, and Ellen Sidransky*

Clinical Neuroscience Branch, National Institute of Mental Health, National Institutes of Health, Bethesda, Maryland

Massive splenomegaly is a frequent finding in patients with Gaucher disease, the most common of the sphingolipidoses. Even so, the risk for splenic rupture and intracapsular hemorrhage has not been emphasized due to the rarity of this occurrence and the fibrotic, rubbery consistency of splenic tissue in these patients. We report two adult patients with type 1 Gaucher disease who suffered life-threatening splenic bleeds that were not acutely diagnosed. Both patients ultimately required emergent splenectomies. Factors complicating the diagnosis of splenic hemorrhage in patients with Gaucher disease are discussed. Am. J. Hematol. 64:140–142, 2000. Published 2000 Wiley-Liss, Inc.†

Key words: glucocerebrosidase; Gaucher disease; splenectomy; splenic hemorrhage

INTRODUCTION

Splenomegaly is a common finding in patients with all three types of Gaucher disease, the inherited deficiency of the enzyme glucocerebrosidase. Individuals with type 1 (non-neuronopathic) Gaucher disease may develop hepatosplenomegaly, thrombocytopenia, anemia, and bone disease, although they lack the neurologic involvement seen in patients with type 2 and type 3 disease. The enormously enlarged spleens of some affected patients usually result in grossly protuberant abdomens which can cause young children to have difficulty ambulating and respiratory compromise. Moreover, hypersplenism in these patients contributes to thrombocytopenia, leading to easy bleeding and bruising. Even so, the risk for splenic injury, including rupture and intracapsular hemorrhage, has not been completely evaluated. In fact, it has been suggested that spleens in patients with Gaucher disease may be less prone to rupture because the splenic tissue is more fibrotic and of tougher consistency [1]. We report two adult patients with type 1 Gaucher disease who suffered splenic bleeds that were not initially diagnosed. In both cases, the precipitating trauma was either mild or unrecognized.

PATIENT HISTORIES

Case 1

This 30-year-old man of European ancestry (genotype N370S/E326K + c.203-204insC) was diagnosed with Gaucher disease as a child when he presented with moderate hepatosplenomegaly. He lived a vigorous life in Alaska, where he was employed as a kayaking guide and carpenter and had not sought medical care for several years. During an interval follow-up evaluation in our clinic, he reported a recent history of constipation and gassy abdominal pains. He recounted that two weeks earlier, during a period of hard labor as a construction worker, he had felt ill and lost consciousness, awakened, vomited, and lost consciousness again. There was no history of trauma. He was evaluated in an emergency room, found to have a hematocrit of 33.0% and was given intravenous fluids for dehydration. On examination in our clinic, he had a soft abdomen with positive bowel sounds, no peritoneal signs, an enlarged spleen (19 cm below the costal margin) and liver (4–5 cm below the costal margin), a normal rectal exam, and mild tenderness over the left lower quadrant. Ambulation resulted in some discomfort. Laboratory studies showed a hematocrit of 25.8%, a platelet count of 113,000, an elevated total bilirubin of 2.1 (normal range 0.1–1.0) with 0.3 direct (normal 0–0.2), and 1–5 RBC/hpf on urinalysis.

*Correspondence to: Ellen Sidransky, M.D., Clinical Neuroscience Branch, NIMH, Bldg. 49, Room B1EE16, 49 Convent Drive, MSC4405, Bethesda, MD 20892-4405.
E-mail: sidranse@irp.nimh.gov

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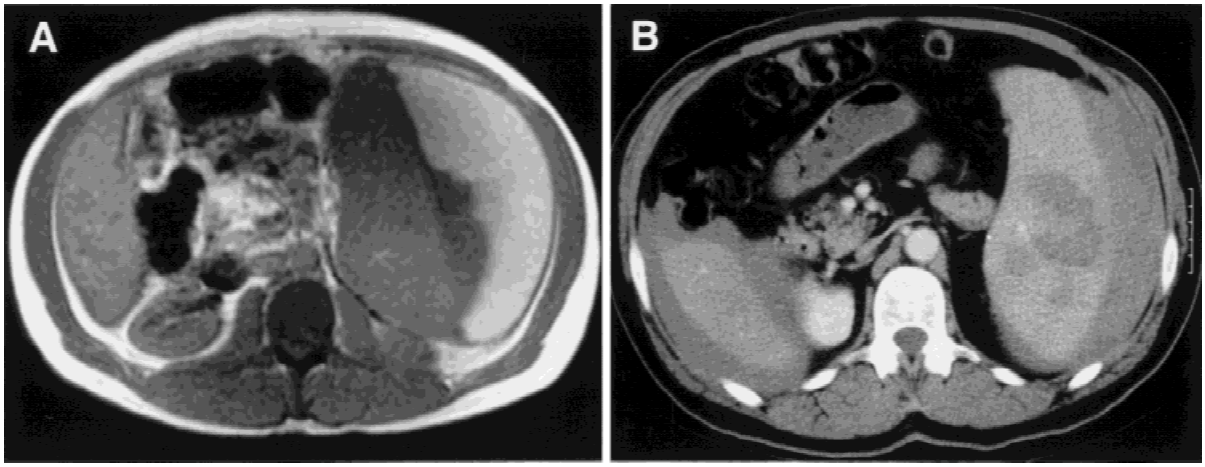

Fig. 1. (A) T1-weighted spin echo MRI image of the upper abdomen of case 1 demonstrating an enlarged spleen with a large hyperintense subcapsular hematoma. (B) CT scan of the upper abdomen of case 2 demonstrating a subcapsular splenic collection of blood, with hemorrhage also seen around the tip of the liver.

An abdominal MRI, performed as a routine follow-up study, revealed a large, subcapsular splenic bleed with free blood in the pelvis (Fig. 1A). He underwent an emergent splenectomy, and a 2,225 gram spleen was removed. This organ measured 30 × 17 × 8 cm, and a recent, massive hematoma with a thin capsule was noted.

Case 2

This Ashkenazi man (genotype N370S/N370S) was diagnosed with Gaucher disease at 40 years of age when he was noted to have thrombocytopenia. Almost twenty years later, en route to the airport, he slipped and fell on ice, injuring his left side. He did not consider this injury to be serious and continued his transcontinental journey. However, two weeks later, he collapsed while at work. He was taken to an emergency room where a CT evaluation revealed a ruptured spleen with a hemoperitoneum and three fractured ribs (Fig. 1B). Laboratory studies showed a hematocrit of 34.1% and a platelet count of 66,000. A splenectomy was performed, and a 1,950 gram spleen was removed. The spleen measured 25 × 18 cm in its greatest diameter, and the lateral surface was covered by freshly clotted blood. A 9 × 4 cm parenchymal hematoma was noted within the inferior half of the spleen communicating with the rupture site on the lateral surface. On the medial surface an 8 cm diameter cystic hematoma was also found immediately deep to the capsule.

Our review of the literature revealed another case, this time of a 21-year-old patient from Italy who was diagnosed with Gaucher disease after he ruptured his spleen by falling from a low stepladder [2]. Several days after his fall, he became hypotensive, and a splenectomy was performed. Pathologic studies showed numerous Gaucher cells in the spleen and led to the diagnosis.

DISCUSSION

These cases illustrate that patients with Gaucher disease and splenomegaly are vulnerable to splenic injury. Although splenic rupture in this population is a rare occurrence, it is imperative to make the diagnosis without delay because of the risk for a subsequent life-threatening hemorrhage. Splenic injury may result from pathologic rib fractures which occur with increased frequency in patients with Gaucher disease and often after very mild trauma, i.e., hugging causing “romantic fractures” [3]. The evaluation of Gaucher patients with splenic injury may be complicated, as these individuals often have chronic anemia. Thus acute blood values must be compared with baseline studies, and sequential hematocrits should be performed to allow for equilibration after an acute, intravascular loss of blood. A splenic bleed may be difficult to clinically differentiate from splenic infarction, another common complication of Gaucher disease [4]. Orthostatic blood pressure measurements, serial hemoglobin levels, and an abdominal MRI or CT should be performed in any patient with Gaucher disease, splenomegaly, and acute or persistent abdominal pain or a history of an abdominal or chest injury. Affected patients may present several weeks after the injury, because an enlarged spleen may temporarily tamponade bleeding and the precipitating trauma may not be recalled. An increased awareness of this complication may allow a life-saving splenectomy to be performed promptly.

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